

CLAIMS

We claim:

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1. An isolated or recombinant nucleic acid which hybridizes to a nucleic acid having the sequence set forth in SEQ ID NO:1, 3 or 5.
  2. A nucleic acid comprising the sequence set forth in SEQ ID NO:1, 3 or 5.
  3. The isolated or recombinant nucleic acid of claim 1 or 2, wherein said nucleic acid comprises or hybridizes to a sequence selected from the group consisting of nucleotides 251 to 769 of SEQ ID NO:3, nucleotides 788 to 1822 of SEQ ID NO:3, nucleotides 1901 to 2018 of SEQ ID NO:3, nucleotides 2189 to 2320 of SEQ ID NO:3 and nucleotides 2330 to 2356 of SEQ ID NO:3.
  4. An isolated or recombinant polypeptide having the amino acid sequence of SEQ ID NO:2, 4, 6, 7, 8 or 9, wherein said polypeptide is a proteoglycan.
  5. The isolated or recombinant polypeptide of claim 4, wherein said proteoglycan is an extracellular matrix protein, plasma membrane associated protein, plasma membrane protein, cytoplasmic protein and/or intranuclear protein.
  6. The polypeptide of claim 4, wherein said polypeptide is a component of the interphotoreceptor matrix.
  7. The polypeptide of claim 4, wherein an abnormality in the amino acid sequence, activity, function or regulation of said polypeptide is associated with a disease or abnormality.
  8. The polypeptide of claim 7, wherein said disease or abnormality is associated with abnormal IPMC activity.

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9. The polypeptide of claim 7, wherein said disease or abnormality is an ocular disease.
10. The polypeptide of claim 7, wherein said disease or abnormality is a non-ocular or systemic disease or abnormality.
11. The polypeptide of claim 9, wherein said disease or abnormality is selected from the group consisting of a retinal detachment chorioretinal degeneration, retinal degeneration, macular degeneration, photoreceptor degeneration, RPE degeneration, cone degeneration, mucopolysaccharidosis, rod-cone dystrophy and cone-rod dystrophy.
12. The polypeptide of claim 11, wherein macular degeneration is age-related macular degeneration.
13. An isolated or recombinant polypeptide comprising an amino acid sequence that diverges no more than 40% from the amino acid sequence represented by SEQ ID NO:2, 4, 6, 7, 8 or 9, wherein said polypeptide comprises an extracellular matrix protein.
14. A kit for detecting mutations in an IPMC gene resulting in a susceptibility to a disease or condition associated with abnormal IPMC activity, said kit comprising at least one oligonucleotide primer capable of differentiating between a normal IPMC gene and an IPMC gene with one or more nucleotide differences.
15. The kit of claim 14, wherein said ocular disease is characterized by abnormal extracellular matrix formation.
16. The kit of claim 15, wherein said extracellular matrix is the interphotoreceptor matrix.

17. The kit of claim 14, wherein said disease or abnormality is an ocular disease selected from the group consisting of a retinal detachment, chorioretinal degeneration, retinal degeneration, macular degeneration, photoreceptor degeneration, RPE degeneration, cone degeneration, mucopolysaccharidosis, rod-cone dystrophy and cone-rod dystrophy.
18. A method for treating or preventing the development of disease in a subject, comprising administering to the subject an effective amount of an IPMC therapeutic.
19. A method of inhibiting photoreceptor death, comprising administering to the subject an effective amount of an IPMC therapeutic.
20. A method for preventing or treating retinal detachment, comprising administering an effective amount of an IPMC therapeutic.
21. The method of claim 18, wherein said IPMC therapeutic is a nucleic acid that comprises all or a portion of the nucleotide sequence set forth in SEQ ID NO:1, 3 or 5.
22. The method of claim 18, wherein said IPMC therapeutic is a polypeptide that comprises all or a portion of the amino acid sequence set forth in SEQ ID NO:2, 4 or 6.
23. A method for identifying agents capable of binding to all or a portion of an IPMC amino acid sequence, comprising:
- (1) incubating said IPMC protein with a composition comprising said agent under conditions that permit binding of said agent to said IPMC protein and thereby creating a complex; and
  - (2) isolating said IPMC binding protein from said complex.
24. A method for identifying agents capable of binding to all or a portion of an IPMC nucleic acid sequence, comprising:

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- (1) incubating said IPMC nucleic acid with a composition comprising said agent under conditions that permit binding of said agent to said IPMC nucleic acid and thereby creating a complex; and
  - (2) isolating said IPMC nucleic acid binding protein from said complex.
25. A method for identifying a compound capable of modulating IPMC gene expression in a cell, comprising:
- (1) incubating a cell with said compound under conditions that permit said compound to exert a detectable regulatory influence over an IPMC gene, thereby altering IPMC gene expression; and
  - (2) detecting an alteration in said IPMC gene expression.
26. An antibody capable of specifically binding to all or a portion of an IPMC protein sequence set forth in SEQ ID NO:2, 4 or 6.
27. The antibody of claim 26, wherein said antibody is an autoantibody.
28. A method for establishing an IPMC genetic population profile in a population of individuals having ocular disease or abnormality, comprising determining the IPMC genetic profile of the individuals in the population and establishing a relationship between IPMC genetic profiles and the phenotype of the individuals.
29. The method of claim 28, wherein said ocular disease or disorder is selected from the group consisting of retinal detachment, chorioretinal degeneration, macular degeneration, photoreceptor degeneration, RPE degeneration, cone degeneration, mucopolysaccharidosis VII, rod-cone dystrophy or cone-rod dystrophy.
30. A method for selecting the appropriate IPMC therapeutic to administer to a subject having an ocular disease or abnormality, comprising determining the IPMC genetic profile of an individual and comparing the individual's IPMC genetic profile to an

IPMC genetic population profile, ~~thereby~~ selecting the appropriate IPMC therapeutic for administration to the subject.

31. A pair of single-stranded DNA primers for amplification of an IPMC nucleic acid sequence by polymerase chain reaction, wherein the sequence of said primers is capable of generating a PCR product when provided to a nucleic acid template and subjected to PCR reaction conditions, wherein the use of said primers in an amplification reaction results in the synthesis of DNA having no less than 40% identity with all or a portion of the sequence represented by SEQ ID NO:1, 3 or 5.
32. The DNA primers of claim 31, wherein said primers comprise primer pairs that have the sequences set forth in SEQ ID NOs:10 and 11, SEQ ID NOs:12 and 13, SEQ ID NOs:14 and 15, SEQ ID NOs:16 and 17, SEQ ID NOs:18 and 19, SEQ ID NOs:20 and 21, SEQ ID NOs:22 and 23, SEQ ID NOs:24 and 25, SEQ ID NOs:26 and 27, SEQ ID NOs:28 and 29, SEQ ID NOs:30 and 31, SEQ ID NOs:32 and 33, SEQ ID NOs:34 and 35, SEQ ID NOs:36 and 37, SEQ ID NOs:38 and 39, SEQ ID NOs:40 and 41, SEQ ID NOs:42 and 43, and SEQ ID NOs:44 and 45.

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